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PREVENTION OF BLINDNESS COMMITTEE
of the Union of Counties Associations for the Blind.

REPORT

ON

HEREDITARY BLINDNESS.

General.

1. A large number of investigations into hereditary blindness, made over a long period of years, have been published in scientific journals, both in this country and abroad. A mass of data has been collected by individual ophthalmologists, but the correlation of the data is a matter for the genetic expert who has already done a good deal of work on it ; for example, Dr. Julia Bell has published from the Galton Eugenics Laboratory, University College, the results of her detailed investigations on the subject in the Nettleship Memorial Volumes. It is hoped that in the future Public Health Departments may be willing to undertake a certain amount of investigation of cases which come before them in the course of their work, with a view to submitting them to scientific analysis by a genetic expert. Notwithstanding the amount of detailed investigation already carried out, the data are still being obtained from too selected a field, and a great difficulty presents itself at the outset in any attempt to estimate accurately the extent to which blindness, as a whole, could be prevented by the limitation of parenthood in individual cases. This stumbling-block arises from the fact that we have no authentic knowledge concerning the extent to which diseases of genetic origin occur also sporadically, and apparently devoid of all genetic potentiality. We can point to cases in which the marriage of one man or woman suffering from, say, Retinitis Pigmentosa, has resulted in some forty cases of blindness in subsequent generations. All will agree that such a possibility is intolerable, but we have no idea what proportion of the total number of persons suffering from some such disease has married with no ill result to their descendants.

2. The Committee on the Prevention of Blindness have already published a Report dealing with the Certification of Blindness, one of the objects of which is to provide that the occasion of certification should also be utilised for the purpose of obtaining a proper record of the eye condition of every person examined. This form of report will in certain cases indicate the possibility of hereditary disease which requires further investigation, but neither the ophthalmologist nor the specialist in heredity would be able to form an opinion as to the hereditary nature of the disease without a pedigree.

Much is known by medical men of the probability of hereditary transmission of a disease, but not enough to justify the Consultant in giving definite advice against parenthood, unless his opinion on the disease *quâ* disease is reinforced by the evidence of a pedigree. It is essential that such a pedigree should contain relevant information, and should apply, as strictly as possible, the principles set out in Appendix I.

The records obtained through the use of the form of report, including a pedigree, should furnish a mass of data which will give a measure of the extent of the hereditary causation of blindness, and should enable reliable conclusions to be based, as they would be for the first time, on a fair sample. Any observations made in

this Report must be taken as provisional to this extent, that they may require modification or amplification in the light of the further data which we hope will be obtained through the certification reports referred to.

3. It will be seen, therefore, that it is not possible to-day to make any accurate estimate of the proportion of the total amount of blindness due to a particular disease, which is preventable by regulation in individual cases. It is, however, possible for an ophthalmologist, in consultation with a genetic expert, to state for any individual, whose family history has been carefully investigated, the degree of risk of blindness in succeeding generations which parenthood would involve.

4. In the light of the foregoing paragraphs, it will be evident that any generalisations on the subject of hereditary blindness are likely to be vague or even misleading. It is doubtful whether the incidence of hereditary blindness would be immediately affected to any marked degree merely by preventing blind couples from having children. Hereditary blindness, in common with other inherited tendencies, is inherited not only through persons actually suffering from it, but also (and in some diseases predominantly) through persons who, though not themselves affected, are, nevertheless, carriers of the taint. Abstention from parenthood by blind persons who are the subject of hereditary eye disease is not therefore of itself sufficient.

An inter-marriage of blind persons should always be approached with caution, and it may be said that parenthood of persons suffering from inheritable blindness is inadvisable unless the ophthalmologist is prepared to say that the risk is so slight as to be negligible.

Classification of Main Hereditary Diseases.

5. The main hereditary diseases or deformities to be considered may be divided into classes according to the prognosis of the disease or deformity:—

(*a*) Those in which heredity is, in the majority of cases, definitely established, and sight is seriously threatened:—

Retinitis Pigmentosa and Leber's Disease.

Retinitis Pigmentosa may be transmitted to children before the disease has manifested itself in the parent. Many cases labelled as Retinitis Pigmentosa are, in reality, Syphilitic Choroido-retinitis, so that statistics with regard to this disease require the most careful scrutiny.

Leber's Disease. In some cases the ultimate vision, permanently retained, is not worse than 6/24, and good fields of vision are preserved. This disease mainly attacks males, but it is only transmitted through females, who rarely suffer from the disease themselves. Exceptional cases, however, are known in which the transmission is through males.

The Optic Atrophy in an hereditary form which is cited by some writers does not appear as an entity apart from Leber's Disease.

(*b*) Those in which heredity is, in many cases, definitely established, but which would not, in all probability, render the patient certifiable as blind, or are amenable to treatment:—

Albinism, Aniridia, Cataract (Congenital and Lamellar), Coloboma of the Iris, Congenital Night-blindness, Ectopia Lentis, Familial Degeneration of the Cornea, Glioma of the Retina.

Some of the foregoing are rare. Buphthalmos falls within this category, but only a slight hereditary tendency can be proved.

Aniridia and Coloboma of the Iris may appear in various members of a family, but only in exceptional instances can be regarded as causes of certifiable blindness.

Buphthalmos. This may be improved by operation, while some cases undergo a natural cure.

Cataract is an operable disease, with resulting improvement in vision.

Glioma is monocular in many cases, and life is preserved and vision in the unaffected eye kept intact by sufficiently early removal of the affected eye. Further organised research may, however, reveal that there is an hereditary type of Glioma which tends to be bilateral.

(*c*) Those in which heredity is, in many cases, definitely established, but which are of very rare occurrence, and may cause partial or complete blindness according to the degree of the disorder:—

Anophthalmos, Familial Macular Degeneration, Microphthalmos, Oxycephaly (with consecutive optic atrophy).

Oxycephaly. More than one child in a family may be affected, but at present there is no reliable evidence as to heredity.

Myopia.

6. The fact that liability to myopia may be accentuated in the children of myopic parents is admitted ; but that the risk of blindness is greater in this special class of myopes than in that class whose parents are not myopic cannot be said to be established.

Transmission of Disease : The View of the Eugenist.

7. The transmission of disease may be considered purely from a eugenic point of view, that is to say, from the point of view of the prevention of the transmission of disease, regardless of all other considerations whatsoever. From this point of view, a eugenist would probably urge that ascertained facts would justify the following conclusions. These conclusions, however, aim at the prevention of eye disease, and not necessarily at the prevention of blindness, for, as pointed out in paragraph 5 above, a number of these diseases do not result in blindness at all:—

(*a*) Any man or woman who has had Aniridia, Congenital Cataract, Glioma of the Retina, Hereditary Opacity of the Cornea or Ectopia Lentis, Microphthalmos, Retinitis Pigmentosa, and of whom either parent has been similarly affected, is likely to transmit the defect and should abstain from parenthood.

(*b*) Any normal parents who have more than one child affected with any one of the above-mentioned diseases, or with Buphthalmos or Total Colour-Blindness, should have no more children, and such children as they have should abstain from parenthood—whether they themselves be affected or not. Even if the parenthood of such children does not lead at once to the manifest reproduction of the disease, it inevitably adds to the latent defect in the population, which becomes manifest when two people carrying such latent defect marry and have children.

(*c*) If, in all stocks in which Leber's Disease (Hereditary Optic Atrophy) has occurred more than once, the sisters of affected males were to abstain from parenthood, the disease would be almost exterminated, except for sporadic cases of no genetic significance. In general, there is no risk to the descendants of men who are suffering from Leber's Disease ; the cases in which

they transmit the disease are very rare, but should be borne in mind, and if one such case in a family has occurred, all other affected members of *that* stock should be regarded as liable to transmit.

General Conclusions.

8. The statements made in paragraph 7 are referred to in order to give some indication of the view of the eugenists, but we cannot, as a Committee on the Prevention of Blindness, subscribe to all the limitations of parenthood which are proposed by them, if only for the reason that some of the diseases do not cause blindness.

We are of opinion that there are no data at present available which would warrant any but the most general conclusions, and it seems to us that—

(*a*) Complete statistics, compiled (i) on a uniform basis, (ii) on the evidence of properly qualified persons, and (iii) including all persons affected by the diseases *and not only those selected from certain pedigrees,* would assist in determining the total amount of blindness which can be prevented by control of parentage ; at present such statistics do not exist.

(*b*) Better certification of the blind population over a period, and the ascertainment of the cause of blindness through the use of the form of report and certificate which the Committee has recommended (of which a revised copy with special provision for recording facts in cases which are of genetic origin appears as Appendix II) should provide data on which to found more accurate conclusions than can be made on existing evidence.

Nevertheless, there are certain general conclusions which we think may be drawn with reasonable confidence prior to the collection of the information referred to at (*a*) and (*b*) above. These are:—

(*c*) From the point of view of the causation of blindness, the most important inheritable diseases which have to be considered are Retinitis Pigmentosa and Leber's Disease (Hereditary Optic Atrophy).

(*d*) If a member of a stock, in which there is definitely inheritable disease causing blindness, even if the disease is not manifest in his or her case, marries, he or she should be strongly urged to consult an ophthalmologist who, with the assistance of a pedigree, would advise as to abstention from parenthood. This statement need not apply to males in a stock carrying Leber's Disease, except in the very exceptional families in which males have been known to transmit.

(*e*) Unless there is medical evidence to show that the case does not fall within the hereditary class, a blind person contemplating marriage should seek the advice of a competent ophthalmologist before marriage, in view of the complex nature of the problem and the serious handicap imposed upon the children of persons suffering from inheritable eye disease. The ophthalmologist will no doubt obtain a pedigree and, if necessary, consult a genetic expert.

In conclusion, we think it right to point out that we have dealt in this Report solely with the medical aspect of Hereditary Blindness, and not with the social aspect.

A Bibliography of publications relating to Hereditary Eye Disease is printed as Appendix III to this Report.

Signed on behalf of the Prevention of Blindness Committee,

P. M. EVANS,
Chairman.

23*rd April,* 1933.

PREVENTION OF BLINDNESS COMMITTEE

of the Union of Counties Associations for the Blind.

APPENDIX I.

Particulars Required for drawing up a Pedigree.

General Note.

Where hereditary defects other than blindness or defective sight, such as deafness, epilepsy, mental deficiency, hare lip or cleft palate, &c., appear in any member of the family, this should be noted.

Where there are cases of half-brothers or sisters in any of the following categories, a note similar to the note regarding full blood relations is desirable.

Persons.	*Particulars.*
1. NAME OF PATIENT.	Clinical description of eye conditions. Sex. Age. Age at onset of eye trouble. Consanguinity of parents ; if not known, *state*.
(*a*) Number of brothers.	Age of each. Whether suffering from eye trouble, or *not*. Clinical description of eye conditions, if manifested, and age at onset. Any other defect.
(*b*) If married, children of each brother.	Number of each sex. Which suffered from eye trouble (sex). Clinical description if possible, and age at onset. Any other defect.
(*c*) Number of sisters.	Age of each. Whether suffering from eye trouble, or *not*. Clinical description of eye conditions, if manifested, and age at onset. Any other defect.
(*d*) If married, children of each sister.	Number of each sex. Which suffered from eye trouble (sex). Clinical description if possible, and age at onset. Any other defect.
2. PATIENT'S MOTHER.	Age. Whether suffering from eye trouble, or *not*. Clinical description of eye conditions, if manifested, and age at onset. Any other defect.

Persons.	*Particulars.*
(*a*) Mother's brothers. (Maternal uncles.)	Number. How many suffering from eye trouble, or *not.* Clinical description of eye conditions, if manifested, and age at onset. Any other defect.
(*b*) Mother's brothers' children. (Each brother's family separately.) (Maternal cousins.)	Number of each sex. Which suffered from eye trouble (sex). Clinical description if possible, and age at onset. Any other defect.
(*c*) Mother's sisters. (Maternal aunts.)	Number. How many suffer from eye trouble, or *not.* Clinical description of eye conditions, if manifested, and age at onset. Any other defect.
(*d*) Mother's sisters' children. (Each sister's family separately.) (Maternal cousins.)	Number of each sex. Which suffered from eye trouble (sex). Clinical description if possible, and age at onset. Any other defect.
3. Patient's Mother's Parents. (Maternal grandparents.)	State if consanguineous, or if *not known.* Which suffered from eye trouble. Clinical description of eye conditions, and age at onset.
4. Patient's Father.	Age. Whether suffering from eye trouble, or *not.* Clinical description of eye conditions, if manifested, and age at onset. Any other defect.
(*a*) Father's brothers. (Paternal uncles.)	Number. How many suffering from eye trouble, or *not.* Clinical description of eye conditions, if manifested, and age at onset.
(*b*) Father's brother's children. (Each brother's family separately.) (Paternal cousins.)	Number of each sex. Which suffered from eye trouble (sex). Clinical description if possible, and age at onset. Any other defect.
(*c*) Father's sisters. (Paternal aunts.)	Number. How many suffer from eye trouble, or *not.* Clinical description of eye conditions, if manifested, and age at onset.
(*d*) Father's sisters' children. (Each sister's family separately.) (Paternal cousins.)	Number of each sex. Which suffered from eye trouble (sex). Clinical description if possible, and age at onset. Any other defect.
5. Patient's Father's Parents. (Paternal grandparents.)	State if consanguineous, or *not known.* Which suffered from eye trouble. Clinical description of eye conditions, and age at onset.
6. Patient's Children (if any).	Number of each sex. Which suffered from eye trouble (sex). Clinical description if possible, and age at onset. Any other defect.

LOCAL AUTHORITY.

STRICTLY PRIVATE AND CONFIDENTIAL.

Examination for Admission to Register of Blind Persons.

Name of Person Examined..... *Sex* .....

Occupation (*Specify, with dates*)..... *Date of Birth*.....

Previous Occupations (*Specify, with dates*).....

Address .....

EXAMINATION OF EYES.

NOTE.—The answers to questions in Sections 2 and 3 are required in order that reliable information may be obtained as to the causes of blindness with a view to assisting in devising appropriate measures for its prevention. It is therefore of importance that great care should be taken in making these replies as accurate as possible, and that entries should be made for each eye.

This section should be completed before forwarding to the Ophthalmic Surgeon.

It is intended that Section 2 should be confined to a record of what the ophthalmologist actually finds in examining the eyes, and Section 3 should state as far as possible the causes to which these conditions are attributable, *e.g.*, if the examination shows interstitial keratitis caused by syphilis "interstitial keratitis" should be entered under "Cornea" in Section 2 (*e*), and "syphilis" under Section 3 B (3).

The findings in Sections 2 and 3 should be entered in Section 6.

1.—HISTORY OF CASE.

	Right Eye.	Left Eye.
(*a*) Age at onset of eye trouble which led to blindness	.....	.....
(*b*) Age at onset of blindness	.....	.....

2.—PHYSICAL EXAMINATION OF THE EYES.

(*a*) Visual Acuity—

	Right Eye.	Left Eye.
(i.) Unaided	.....	.....
(ii.) With focus properly corrected	.....	.....

(iii.) Both eyes together unaided.....

(iv.) Both eyes together with focus properly corrected.....

(*b*) Generally age at which applicant was unable to follow usual occupation on account of eye trouble.

	*Right Eye.	*Left Eye.
(*b*) Field of vision	(i.) Good	(i.) Good
* (*Please underline appropriate condition.*)	(ii.) Contracted	(ii.) Contracted
	(iii.) Less than 10°	(iii.) Less than 10°
	(iv.) Central Scotoma	(iv.) Central Scotoma
	(v.) Hemianopia	(v.) Hemianopia
	(vi.) Nil	(vi.) Nil
(*c*) Globe (tension, muscles, *e.g.*, squint, nystagmus, &c.)	.....	.....
(*d*) Lids	.....	.....
(*e*) Cornea	.....	.....
(*f*) Iris	.....	.....
(*g*) Ciliary body	.....	.....
(*h*) Lens	.....	.....
(*j*) Vitreous	.....	.....
(*k*) Retina	.....	.....
(*l*) Choroid	.....	.....
(*m*) Optic nerve	.....	.....

(*n*) Prognosis.....

(*o*) Is there any reason to suspect malingering? .....

(*p*) Is re-examination for any reason desirable? If so, when? .....

3.—CAUSES OF EYE DEFECTS (as ascertained in 2, see marginal note).

The appropriate cause or causes of the condition found in Section 2 should be underlined in this Section 3, indicating for each eye the main cause with the figure (1), and other cause, if any, with the figure (2).

	Right Eye.	Left Eye.
A.—Congenital and Undetermined Causes—		
(1) Congenital, hereditary and developmental defects		
(2) Myopic error		
(3) Other errors of refraction. (Specify)		
(4) Glaucoma, primary		
(5) Cataract, primary		
(6) Other primary ocular defects, *e.g.*, primary detachment of retina. (Specify)		
(7) Tumour—intra-ocular		
(8) Tumour—adnexa		
(9) Other, *e.g.*, amblyopia ex anopsia, &c. (Specify)		
B.—Infectious and Bacterial—		
(1) Ophthalmia Neonatorum		
(2) Gonorrhœa		
(3) Syphilis		
(*a*) Congenital		
(*b*) Acquired		
(4) Trachoma		
(5) Local infection of coats of eye		
(6) Specific Fevers, *e.g.*, smallpox, measles, scarlet, diphtheria. (Specify)		
(7) Meningitis—cerebro-spinal and other non-tuberculous forms. (Specify)		
(8) Tuberculosis		
(9) Eczematous kerato-conjunctivitis, *e.g.*, phlyctenular, &c., not definitely tuberculous		
(10) Septicæmia, acute		
(11) Septicæmia, chronic; auto-toxic, focal sepsis		
(12) Other infections and bacterial diseases. (Specify)		
C.—Traumatic and Chemical—		
(1) Industrial—		
(*a*) Trauma. (State precisely)		
Cause		
Trade		
Process		
(*b*) Industrial disease. (State precisely)		
Cause		
Trade		
Process		
(2) Non-industrial—		
(*a*) Trauma. (State precise cause)		
(*b*) Systemic poisoning, *e.g.*, tobacco, alcohol, lead, arsenic, thallium, &c. (State precise cause)		
(*c*) Birth trauma		
(*d*) War trauma		
(3) Indirect: sympathetic ophthalmia		
(4) Trauma (category not ascertainable)		
D.—General Diseases—		
(1) Anæmia and other blood diseases. (Specify)		
(2) Vascular diseases, including cerebral vascular lesions. (Specify)		
(3) Diseases of the central nervous system (except meningitis and those mentioned at (4) and (5) below). (Specify)		
(4) Diseases connected with disorders of pregnancy and childbirth, *e.g.*, eclampsia. (Specify)		
(5) Intracranial neoplasm		
(6) Diabetes		
7) Nephritis		
(8) Other general diseases. (Specify)		

It is hoped that Ophthalmic Surgeons will complete as much of this Section as may be possible with the means at their disposal

E.—No information obtainable.

4.—REMARKS, *e.g.*, Family history, &c.

[t is specially desirable that in cases where it is applicable (Leber's Disease, Tay-Sach's Disease, Retinitis Pigmentosa, and Allied Diseases, High Myopia, etc.) as much information as possible regarding relatives who are blind or who have seriously defective vision should be detailed. In all cases of disease which may be of genetic origin it is particularly requested that a definite statement shall be made as to whether or no any relatives have been known to be similarly affected, and whether the parents of the individual were or were not consanguineous. This space should, if possible, not be left blank.

(Please UNDERLINE the words which apply in this Section 5.)

It is hoped that Ophthalmic Surgeons will complete as much of this Section as may be possible with the means at their disposal

5.—OTHER MENTAL, NERVOUS OR PHYSICAL DISABILITY (IF ANY) IN ADDITION TO BLINDNESS, e.g.—

(1) Mental defect.
(2) Epilepsy.
(3) Deafness.
(4) Deaf-mutism.
(5) Crippling defects, bones, joints, spinal, etc. (Specify) .....
(6) Other. (Specify) .....

Enter in this Section the appropriate letters and numbers from Sections 2 and 3, *e.g.*, In a case of Retinitis with Optic Atrophy the certifying Ophthalmic Surgeon might underline in Section 2 (*b*) Field of Vision ii. Contracted, also (*k*) Retina and (*m*) Optic Nerve, and complete Section 6 (A) thus :—

Eye defects found :
(1) *k* (1) *k*
(2) *m* (2) *m*
(3) *b* ii (3) *b* ii

The appropriate letters and numbers from Section 3 will be entered in Section 6 (B)

6.—SUMMARY OF CASE.

			Right Eye.	Left Eye.
A.—Eye defects found (*see* Section 2).	Main	(1)	(1)	(1)
	Other	(2)	(2)	(2)
		(3)	(3)	(3)
B.—Cause of eye defects (*see* Section 3).	Main	(1)	(1)	(1)
	Other	(2)	(2)	(2)

(Please UNDERLINE the words which apply in this Section 7.)

7.—TREATMENT.

A.—History of Institutional Treatment and Operations for ophthalmic condition or its cause—

(1) Not known.
(2) None.
(3) Medical, successful.
(4) ,, unsuccessful.
(5) Medical, uncompleted.
(6) Surgical, successful.
(7) Surgical, unsuccessful.
(8) ,, uncompleted.

B.—History of Anti-Syphilitic Treatment—

(1) Not known.
(2) Regular course at hospital or clinic.
(3) By private practitioner.
(4) Other treatment, if any. (Specify).....
(5) No medical treatment.

C.—Treatment recommended—

(1) None. (2) Medical. (3) Surgical. (4) Optical. (5) Educational, *i.e.*, by special methods (if under 16).

D.—Would Treatment probably remove person from blind category?

(1) Yes. (2) No. (3) Doubtful. (4) Yes, but inadvisable on general grounds.

E.—Is Wasserman or other serological test recommended?

(1) Yes. (2) No. (3) Result (when known).

[*If the applicant has already had a serological test carried out, UNDERLINE* (4) *or* (5) *whichever is appropriate.*]

(4) Positive. (5) Negative.

CERTIFICATE.

(The answers to the following questions must be YES or NO).

(1) Do you consider the person "so blind as to be unable to perform any work for which eyesight is essential" within the meaning of the Blind Persons Act, 1920? (*See* Note A) ... ...

(2) If the person is a child under 16, is it blind within the meaning of Section 69 of the Education Act, 1921, namely, "too blind to be able to read the ordinary school books used by children," *i.e.*, is the child either—

(*a*) Totally blind, or so blind that it can only be appropriately taught in a Special School or Class for **blind** children? or .....

(*b*) With such power of vision that, though it cannot read ordinary school books or cannot read without injury to its eyesight, it can appropriately be taught in a Special School or Class for the **partially blind**? (*See* Note B) ....

(3) If the person is a child under 16, do you consider it likely that the child will be blind within the meaning of the Blind Persons Act, 1920, on attaining the age of 16? ... ...

(4) If the person is 16 years of age or over and not at present blind within the meaning of the Blind Persons Act, 1920, do you consider it likely that the person will become blind within the meaning of the Act during the next 4 years? (*See* Note C) ... .

Address of place of Examination.....

.....

Signature.....

Certifying Ophthalmic Surgeon.

Date.....19

NOTES.

(A.) Blindness within the meaning of the Blind Persons Act, 1920, means "so blind as to be unable to perform any work for which eyesight is essential" (*See* Section I. of the Blind Persons Act, 1920). The Ministry of Health has explained the criteria which should be adopted in interpreting this definition in Circular 681 of the 29th March, 1926, and Circular 780 of the 27th April, 1927.

(B.) It must be understood that children who, by means of suitable glasses, are able to read the ordinary school books used by children, without fatigue or injury to their vision, do not fall within the above categories. Children included in (2) (*b*) above must not be entered on the register of blind persons.

(C.) The primary object of this question is to determine whether a person not yet blind should be trained in anticipation of his becoming blind by the time the training is complete.

APPENDIX III.

HEREDITARY BLINDNESS.

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